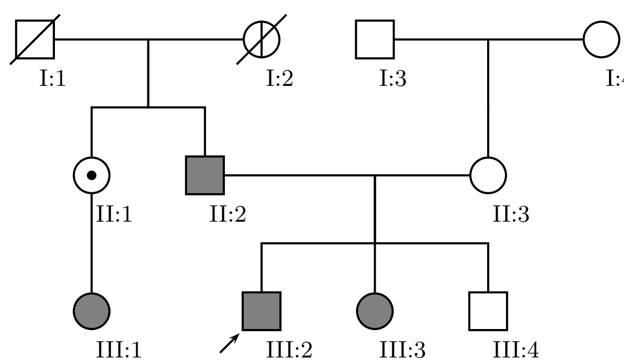
A Program For Automatic Pedigree Construction With pst-pdgr
User Manual and Algorithm Description

Boris Veytsman, borisv@lk.net   Leila Akhmadeeva

March 2012

Abstract

The set of macros in pst-pdgr package allows to typeset complex pedigrees. However, a manual placement of pedigree symbols on a canvas is a time-consuming task. This program produces TEX files from spreadsheets with the data on inheritance for a large class of pedigrees. It has a simple interface and can be used for quite complex pedigrees.



Contents

I User Manual 4

1 Introduction 4

2 Installation 4
2.1 System Requirements . . . 4
2.2 Unix/Linux Installation . . . 4
2.3 Installation in Other Systems . . . 5

3 Configuration 5
3.1 Configuration Variables and Location of Configuration File . . . 5
3.2 Configuration File Format . . . 6
3.3 TEX Output Setup . . . 6
3.4 What to Print . . . 7
3.5 Language and Encoding . . . 8
3.6 Fonts . . . 8
3.7 Lengths . . . 9
3.8 Scaling and Rotation . . . 9

4 Running the Program 10
4.1 Program Invocation And Options . . . 10
4.2 Data File . . . 11
4.3 Twins . . . 13
4.4 Abortions . . . 13
4.5 Childlessness and Infertility . . . 13
4.6 Ordering Siblings and Marriage Partners . . . 19
4.7 Consanguinic Unions . . . 26
4.8 Language-Dependent Keywords . . . 26

II Algorithm Description 29

5 Introduction 29

6 Main Algorithm 29

7 Algorithm for Sorting Siblings and Marriage Partners 30

8 Modifications for Consangunic Unions 31

9 Conclusion 31

10 Acknowledgements 32

List of Figures

1 Example of the Typeset Pedigree in English (Data File from Listing 7) . . . 15
2 Example of the Typeset Pedigree in Russian (Data File from Listing 7) . . . 16
3 Example of a Pedigree with Twins (Data File from Listing 8) . . 17
4 Example of a Pedigree with Abortions (Data File from Listing 9) 18
5 Example of a Pedigree with Childlessness (Data File from Listing 10) . . . 20
6 Pedigree from Listing 12 . . . 22
7 Pedigree from Listing 12 . . . 23
8 Pedigree from Listing 13 . . . 24
9 Pedigree from Listing 14 . . . 25
10 Pedigree from Listing 15 . . . 27
11 Subpedigrees and Downward Tree . . . 30

List of Tables

1 Keywords in Different Languages . . . 28

List of Listings

1 Configuration File: Setting TeX Output . . . 7
2 Configuration File: Choosing Fields to Print . . . 8
3 Configuration File: Choosing Language and Encoding . . . 9
4 Configuration File: Choosing Fonts . . . 9
5 Configuration File: Choosing Lengths . . . 10
6 Configuration File: Choosing Scaling and Rotation . . . 11
7 Examples of Data Files (English and Russian) . . . 14
8 Example of Data File with Twins . . . 17
9 Example of Data File with Abortions . . . 18
10 Example of Data File with Childlessness . . . 19
11 A Data File with a Sorting Problem . . . 21
12 First Solution to the Problem in Listing 11 . . . 21
13 Second Solution to the Problem in Listing 11 . . . 23
14 A Pedigree with Unavoidable Self-Intersections . . . 24
15 A Pedigree with Consanguinic Unions . . . 26

Part I
User Manual

1 Introduction

Medical pedigree is a very important tool for clinicians, genetic researchers and educators. As stated in [1], "The construction of an accurate family pedigree is a fundamental component of a clinical genetic evaluation and of human genetic research." The package `pst-pdgr` [2] provides a set of PSTricks macros (see [3]) to typeset pedigrees. In the framework of `pst-pdgr` the user manually chooses coordinates for each pedigree node on the diagram. While this is relatively easy for small pedigrees, this task becomes increasingly time-consuming for larger ones. There may be several approaches to automate it. For example, one may have data about the patients and their families in a spreadsheet or database. Then it would be useful to generate pedigrees from such data. This is the aim of the program `pedigree` described in this manual.

Spreadsheets and databases can export the data as separated values files ("csv" files for Comma Separated Values). Our program reads these files and outputs LaTeX code with `pst-pdgr` macros. We tried to make this code readable, so a user might tweak it if necessary.

Of course, manually produced LaTeX code is more versatile than the automatically generated one. There are certain limitations for the program: 1. only persons having common genes with the proband or the "starting person" are included in the pedigree; 2. no adopted children, sperm donors or surrogate mothers are shown on the pedigree; 3. only one disease is shown on the chart; 4. the support for consanguinic unions and inbreeding is rather experimental (see Section 4.7). Subsequent versions of the program may ease some of these limitations.

2 Installation

2.1 System Requirements

The program requires Perl version 5 or newer (it was tested with Perl v5.8.8, but should work with any Perl-5). The LaTeX macros require `pst-pdgr` version 0.3 (July 2007) or newer.

2.2 Unix/Linux Installation

If your system has a working `make` program, which is the usual case for Unix-like environments, the supplied `Makefile` installs the executable `pedigree` in `/usr/local/bin`, the libraries in `/usr/local/lib/site_perl` and the manual pages in `/usr/local/man`. This is done by the usual command `make install`.

Optionally you can install files in the `doc` and `examples` subdirectories in the proper places in your system.

2.3 Installation in Other Systems

If your system does not have make, you need to manually perform the following:

1. Install the executable `pedigree.pl` to the place your system can find it.

2. Install the libraries: `Pedigree.pm`, directory `Pedigree` and all files in it to the Perl search path. The latter is listed in the array `@INC`, which can be checked by the command `perl -V` or its equivalent.

3 Configuration

3.1 Configuration Variables and Location of Configuration File

The program defaults are sufficient for most cases. However, if you want to draw pedigrees in a language other than English, or to tweak the layout of the pedigrees, you need to change the program configuration.

The behavior of the program pedigree is determined by *configuration variables.* There are several sources of configuration variables. They are (in the order of increasing priority):

1. Program defaults.

2. The system configuration file[1] `/etc/pedigree.cfg`. On TEXLive the system coniguration files are `$TEXMFHOME/texmf-config/pedigree/pedigree.cfg` and `$TEXMFLOCAL/pedigree/pedigree.cfg`.

3. User configuration file[2] `$HOME/.pedigreerc`.

4. The file specified by the `-c` option (see Section 4.1).

If a file mentioned in this list does not exists, the program silently[3] continues.

Note that even if a configuration file with higher priority exists, the program reads the files with lower priority first. The former *overrides* the latter, but not precludes it from reading. In other words, if `/etc/pedigree.cfg` defines variables `$foo` and `$bar`, and `$HOME/.pedigreerc` defines `$bar` and `$baz`, the program takes `$foo` from the first file, and `$bar` and `$baz` from the second one.

[1]On Unix-like systems, where `/etc` exists
[2]On Unix-like systems, where `$HOME` exists
[3]Unless `-d` option is selected, see Section 4.1

3.2 Configuration File Format

All configuration files mentioned in Section 3.1, have the same format. They are actually snippets of Perl code, executed by the program pedigree. This means, by the way, that all precautions usually taken with respect to programs and scripts, are relevant for configuration files as well. In particular, it is a bad idea to have world-writable system-wide configuration file `/etc/pedigree.cfg`.

The code in configuration files is very simple, and one does not need to know Perl to edit configuration files. There are several simple rules which are enough to understand these files:

1. All text after `#` to the end of the line is a comments. In particular, the lines starting with `#`, are comment lines.
2. Perl commands must end by semicolon `;`.
3. The commands like

   ```
   $xdist=1.5;
   ```

   or

   ```
   @fieldsforprint=qw(Name DoB);
   ```

   assign values to the variables.
4. Variables starting with `$` are scalars and take numerical or string values. Variables starting with `@` are arrays and take list of values.
5. A backslash in single quotes stands for itself, A backslash in double quotes or inside `<<END...END` construction must be doubled. Compare the commands

   ```
   $foo='\documentclass';
   $bar="\\documentclass";
   ```

6. The last command in the file must be

   ```
   1;
   ```

A number of commented configuration files can be found in the `examples` subdirectory of the distribution.

In the remaining parts of this section we describe the configuration variables in detail.

3.3 TEX Output Setup

A number of variables determine what kind of TEX file is produced. An example of their usage is shown on Listing 1.

```
# Do we want to have a full LaTeX
# file or just a fragment?
#
$fulldoc=1;

# What kind of document do we want
#
$documentheader='\documentclass{article}';

# Define additional packages here
#
$addtopreamble=<<END;
\\usepackage{pst-pdgr}
END

# Do we want to print a legend?
#
$printlegend=1;
```

Listing 1: Configuration File: Setting TEX Output

The variable `$fulldoc` determines whether the program produces a full LATEX file with header and preamble (when `$fulldoc=1`), or just a snippet to be included in a larger document (when `$fulldoc=0`). The default is 1.

The variable `$documentheader` is used when `$fulldoc` is 1. It determines the document class of the resulting LATEX file. The default is `article` class, set by `\documentclass{article}`.

By default the preamble of the LATEX file created when `$fulldoc` is 1, contains only the line `\usepackage{pst-pdgr}` and, if the language chosen is not English (see Section 3.5), the calls of babel and inputenc packages. The variable `$addtopreamble`, if set, may contain any other LATEX code you might wish to add to the preamble.

The variable `$printlegend` determines whether to add legend to the pedigree. The default value is 1, and the legend is printed.

3.4 What to Print

The next groups of configuration variables sets the information to be printed in the legend and on the pedigree. It consists of two arrays: array `@fieldsforlegend` is the list of fields (see Section 4.2) which are included in the legend, and array `@fieldsforchart` is the list of fields to print near each node in the pedigree (Listing 2). Setting `@fieldsforchart` to empty array:

```
@fieldsforchart = ();
```

```
# Fields to include in the legend.
# Delete Name for privacy protection.
#
@fieldsforlegend = qw(Name DoB DoD Comment);

#
# Fields to put at the node.
# Delete Name for privacy protection.
#
@fieldsforchart = qw(Name);
```

Listing 2: Configuration File: Choosing Fields to Print

prevents putting additional information on the pedigrees.

The field names are described in Section 4.2. Note that `AgeAtDeath` is a special field: it is the age at death (or empty) calculated as the difference between the death date and the birth date.

3.5 Language and Encoding

The next group of variables describes the language and encoding of the data file input and the LaTeX output. They are shown in Listing 3. The variable `$language` at present can have one of two values: `english` (the default) or `russian`. If the value is `russian`, the output document preamble includes the line

```
\usepackage[russian]{babel}
```

The variable `$encoding` sets the encoding of the LaTeX file if the language is not English. By default it is `cp1251`, if the language is Russian. Set it to `koi8-r` to choose KOI8 encoding. It is worth to note that the data file and the output LaTeX file are assumed to have the same language and encoding.

If `$language` is not `english`, the program recognizes both English and native names of the fields in the data file (see Section 4.2).

3.6 Fonts

There are two kinds of text on the chart: the text above a node and the text below a node[4]. The fonts for them are set by the variables `$belowtextfont` (by default `\small`) and `$abovetextfont` (by default `\scriptsize`). Any LaTeX font declaration like `\sffamily` or `\itshape` is allowed here. See Listing 4 for an example of usage.

[4]The TeX package [2] also allows to place text at both sides of the node, but the program `pedigree` currently does not use this feature.

```
#
# Language
#
# $language="russian";
$language="english";

#
# Override the encoding
#
# $encoding="koi8-r";
```

Listing 3: Configuration File: Choosing Language and Encoding

```
#
# Fonts for the chart
#
$belowtextfont='\small';
$abovetextfont='\scriptsize';
```

Listing 4: Configuration File: Choosing Fonts

3.7 Lengths

The next group of variables (Listing 5) sets the distances between the key elements of the chart. All lengths are in centimeters (actually, in `units`, are defined in PSTricks [3]).

The variable `$descarmA` sets the length of the first segment of the descent line: from the parent node to the sibs line, as measured from the center of the parent (see [2] for more details). By default it is 0.8.

The variables `$xdist` and `$ydist` set the distances between the nodes along horizontal and vertical axes correspondingly. The default for both is 2.

3.8 Scaling and Rotation

Complex pedigrees might be too large to fit on a page. In this case a scaling and (or) rotation might be necessary to print the chart. Of course, changing the lengths described in Section 3.7 might also help, but the scaling described here also changed the size of the pedigree symbols.

There are three variables controlling the scaling and rotation of pedigrees: `$maxW`, `$maxH` and `$rotate` (see Listing 6). The variables `$maxW` and `$maxH` are the maximal width and height of the chart in centimeters. Setting any of them to zero disables scaling.

```
#
#  descarmA in cm
#
$descarmA = 0.8;

#
# Distances between nodes (in cm)
#
$xdist=2;
$ydist=2;
```

Listing 5: Configuration File: Choosing Lengths

The scaling works as follows. If both height and width of the pedigree are smaller than the limits, no scaling is done. In the other case the chart is scaled while preserving the aspect ratio (by changing the value of `unit`, see [3]) to fit into the limits.

The variable `$rotate` sets the orientation of the chart. If it is `no`, the pedigree is never rotated, while if it `yes`, it is always rotated ninety degrees counterclockwise. If this variable is set to `maybe` (the default), the program compares the scaling for the non-rotated and rotated pedigrees, and chooses the orientation for which the scaling is closer to one.

4 Running the Program

4.1 Program Invocation And Options

The program `pedigree` is a command line program. It reads the data from a text file `input_file` and produces an output file with LaTeX macros. The format of the input file is described in Section 4.2. The program invocation is:

```
pedigree [-c configuration_file] [-d] [-o output_file]
         [-s start] input_file
```

(the square brackets show optional arguments).

All arguments but `input_file` are optional. They are described below.

The option `-c` selects a *configuration file.* The format of the configuration file is described in Section 3.1. If this option is absent, the program uses its own default parameters, or system-wide or user's defaults, as explained in Section 3.1.

The option `-d` selects debugging mode. In this mode a lot of debugging messages are dumped to `stderr`.

The parameter `-o` provides the name of the output file. Both `input_file` and `output_file` can be "-", which means `stdin` for the input and `stdout` for the output. If the parameter `-o` is absent, the program tries to guess the name

```
#
# Maximal width and height of the pedigree in cm.
# Set this to 0 to switch off scaling
#
$maxW = 15;
$maxH = 19;

#
# Whether to rotate the page.  The values are
# 'yes', 'no' and 'maybe'
# If 'maybe' is chosen, the pedigree is rotated
# if this provides better scaling
#
$rotate = 'maybe';
```

Listing 6: Configuration File: Choosing Scaling and Rotation

of the output file from the name of the input file. If the input file is `foo.csv`, the output file will be `foo.tex`. On the other hand, if the input file is `stdin`, the output file is `stdout`.

Usually pedigrees are built starting from the proband[5]. Only the people that share genes with the proband, are shown on the pedigree. However, in some cases, for example when there is no proband, or where there are several probands, it is neccessary to override this default and tell the program from which person to start. This is done using the option `-s`. If it is present, it must be followed by the Id of a person in the data file (see Section 4.2 for the discussion of Id).

The option `-v` is special. The invocation `pedigree -v` outputs the version and license information.

4.2 Data File

The input for the program is a separated values file. Usually such files are called CSV for "comma separated values". However, this program uses the vertical bar ("pipe") `|` as a separator. Each line of this file is a *record.* The lines are separated by pipes into *fields.* Most SQL programs produce such files by default. Spreadsheet programs will make them if you choose "Save As..." option, and select `|` as the field separator, and empty text delimiter. We sometimes will call the records "rows" and the fields "columns" to use the familiar spreadsheet metaphor. Normally each row corresponds to a person in a pedigree. We will call this person *the current person* when describing the fields.

[5]The proband is the first person among the relatives who came to a geneticist; he or she is the primary patient.

The width of the fields may not be the same in all rows (or, in other words, the pipes `|` may be disaligned). We make them aligned in the examples included in this manual just to make the text more readable.

The first line of the data file contains the names of the fields ("column headers"). The fields in the subsequent lines must match the order of the headers. An empty field must be still included (as `||` or `| |`). Otherwise the order of columns is arbitrary as long as it is the same for all rows (i.e. matches the order of "column headers" in the first line).

All fields but `Id` are optional. If the value is empty for all rows, the corresponding column can be dropped. If applicable, the default values for this field will be substituted by the program.

On the other hand the data file can include any additional columns as long as their names do not clash with the names listed below and the special name `AgeAtDeath`. These additional columns can be included in the chart or legend as described in Section 3.4.

Here is the list of columns and explanation of their meaning:

Id: Each line (including the special lines described below) must have a unique `Id`. The `Id` may contain only Latin letters and numbers, and start with a letter.

Name: The name of the person described in the current row. There are also *special names* when the current row describes abortions or infertility. They are described below. The names should not contain "special symbols" like #, $, %, _, ^, etc.

Sex: The gender of a person. This column may have one of two values: `male` or `female`. The empty value corresponds to a person with unknown gender.

DoB: The date of birth for the current person. The format is `YYYY.MM.DD`. If the date of birth is not known, the field may be empty or the keyword `unknown` may be used.

DoD: The date of death for current person. The format is the same as for `DoB`: `YYYY.MM.DD`. If this field is empty, the corresponding person is alive. For deceased persons with an unknown date of death use the keyword `unknown`. Note the subtle difference between the fields `DoB` and `DoD`: an empty value for `DoB` is means "unknown birth date" while for `DoD` it means that there is no date of death at all.

Mother: The `Id` of the mother of the person (or empty).

Father: The `Id` of the father of the person (or empty).

Proband This field can be either `yes` for the probands, or empty (or `no`) for other persons. Note that if a pedigree has no probands or several probands, the program does not know, from which node to start the pedigree. Therefore in this case the option `-s` must be used to explicitly set the `Id` of the starting chart node (see Section 4.1).

Condition: This column can have the values `normal`, `obligatory`, `asymptomatic` or `affected`. If it is empty, the default value `normal` is assumed.

Comment: A comment about the person.

Twins: If the current person has twins, they are listed in this column separated by spaces and (or) commas. See Section 4.3 for more details.

Type: This column is used in certain special cases. For abortions it shows the type of the abortion (Section 4.4), for childless people and marriages it shows the type of childnessness (Section 4.5), and for twins it shows the type of twins (Section 4.3).

SortOrder: This column is used when the algorithm for sorting siblings and unions gives a wrong result, and a manual correction is needed. See Section 4.6 for the explanation and examples.

Examples of data files (in English and Russian) are shown in Listing 7 (the Russian keywords are discussed in Section 4.8).

4.3 Twins

The column `Twins` (see Section 4.3) lists all `Id`s of all twins of the given person. The column `Type` can be used to show the type of the twins. The empty value means polyzygotic twins, `monozygotic` means monozygotic twins, and `qzygotic` is used in the case when the type of twins is under doubt. An example of a data file with twins is shown on Listing 8, and the corresponding pedigree on Figure 3.

4.4 Abortions

Aborted pregnancies are described by a special entry in the data file. The field `Name` has the value `#abortion`; the symbol `#` is used to show that this is a special value. The columns `Sex`, `DoB`, `Mother`, `Father` and `Condition` have the usual meaning. The special column `Type` is either empty or be equal to `sab` for self-abortions.

4.5 Childlessness and Infertility

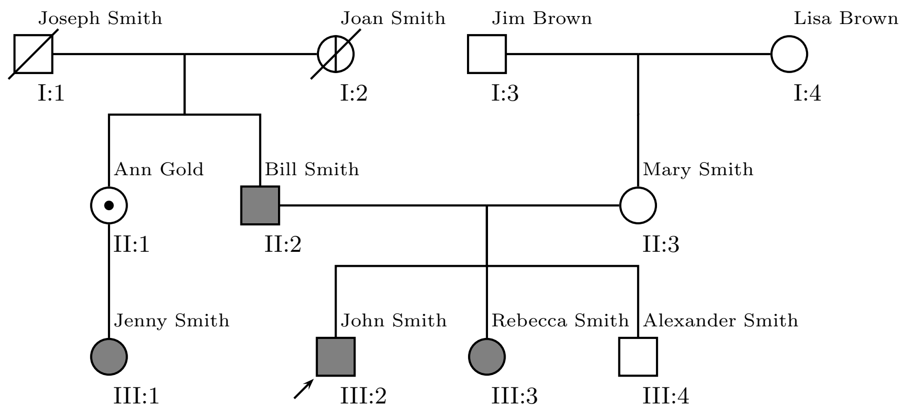
Childlessness is can be a property of a person or a union between two persons. Therefore in this implementation we use a special row rather than a column to report it. As other rows, this one has a unique `Id`. The `Name` column should have a special entry `#childless`. Like `#abortion` (Section 4.4), this special name starts with `#` to distinguish it from "real" names. There are four other columns that have meaning for this row:

Mother: The `Id` of the childless female.

```
Id |Name            |Sex   |DoB       | DoD      |Mother|Father|Proband|Condition |Comment
P  |John Smith      |male  |1970/02/05|          |M1    |F1    | yes   | affected|Evaluated 2005/12/01
M1 |Mary Smith      |female|1940/02/05|          |GM2   |GF2   |       | normal  |
F1 |Bill Smith      |male  |1938/04/03|          |GM1   | GF1  |       |affected |
GM1|Joan Smith      |female|1902/07/01|1975/12/13|      |      |       |asymptomatic
GF1|Joseph Smith    |male  |unknown   |unknown   |      |      |       | normal
GF2|Jim Brown       |male  |1905/11/01|          |      |      |       | normal  |
GM2|Lisa Brown      |female|1910/03/03|          |      |      |       | normal  |
S1 |Rebecca Smith   |female|1972/12/25|          |M1    |F1    |       | affected
S2 |Alexander Smith |male  |1975/11/12|          |M1    |F1    |       | normal
A1 |Ann Gold        |female|1941/09/02|          |GM1   | GF1  |       | obligatory|Aunt of the proband
C1 | Jenny Smith    |female|1969/12/03|          |A1    |      |       | affected | Cousin of the proband
```

```
Идент|ФИО                     |Пол|Рожд      |Умер      |Мать|Отец|Пробанд|Состояние | Комментарий
P    |Иванов Сергей Петрович |муж|1965/08/06|          |M1  |F1  |да     |больн     |
M1   |Иванова Любовь Ивановна|жен|1935/12/01|2005/10/01|    |    |       |норм
F1   |Иванов Петр Ильич      |муж|неизв     |2003/01/25|    |    |       |облигат
S1   |Иванова Анна Петровна  |жен|1968/05/05|          |M1  |F1  |       |норм
K1   |Иванов Иван Сергеевич  |муж|1990/12/01|          |    |P   |       |асимп  |Генетич. иссл. 2005/12/08
K2   |Иванова Дарья Сергеевна|жен|1995/03/24|          |    |P   |       |норм   |Генетич. иссл. 2005/12/08
```

Listing 7: Examples of Data Files (English and Russian)



I:1 Joseph Smith; born: unknown; age at death: unknown.

I:2 Joan Smith; born: 1902/07/01; age at death: 73.

I:3 Jim Brown; born: 1905/11/01.

I:4 Lisa Brown; born: 1910/03/03.

II:1 Ann Gold; born: 1941/09/02; Aunt of the proband.

II:2 Bill Smith; born: 1938/04/03.

II:3 Mary Smith; born: 1940/02/05.

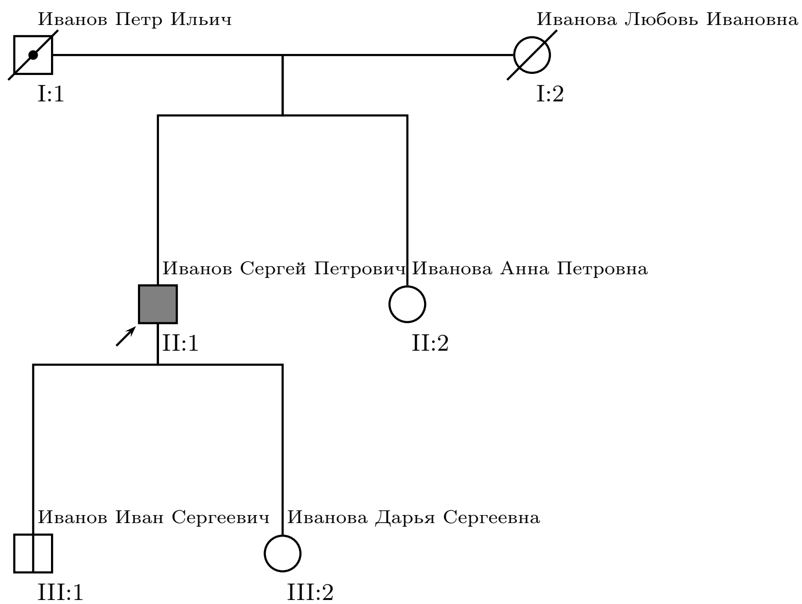
III:1 Jenny Smith; born: 1969/12/03; Cousin of the proband.

III:2 John Smith; born: 1970/02/05; Evaluated 2005/12/01.

III:3 Rebecca Smith; born: 1972/12/25.

III:4 Alexander Smith; born: 1975/11/12.

Figure 1: Example of the Typeset Pedigree in English (Data File from Listing 7)



I:1 Иванов Петр Ильич; род. неизв.; ум. в возр. неизв..

I:2 Иванова Любовь Ивановна; род. 1935/12/01; ум. в возр. 70.

II:1 Иванов Сергей Петрович; род. 1965/08/06.

II:2 Иванова Анна Петровна; род. 1968/05/05.

III:1 Иванов Иван Сергеевич; род. 1990/12/01; Генетич. иссл. 2005/12/08.

III:2 Иванова Дарья Сергеевна; род. 1995/03/24; Генетич. иссл. 2005/12/08.

Figure 2: Example of the Typeset Pedigree in Russian (Data File from Listing 7)

```
Id |Name  |Sex   |DoB       |DoD       |Mother|Father|Proband|Twins|Type
F0 |Adam  |male  |unknown   |unknown   |      |      |       |     |
A0 |Sam   |male  |1950.01.03|unknown   |      |F0    |       | A1  |qzygotic
A1 |John  |male  |1950.01.03|2005.04.12|      |F0    |       | A0  |qzygotic
A2 |Jane  |female|1951.14.15|          |      |      |       |     |
B1 |Jack  |male  |1975.05.06|          |A2    |A1    |       |B2   |monozygotic
B2 |Mike  |male  |1975.05.06|          |A2    |A1    |       |B1   |monozygotic
B3 |Pam   |female|1973.11.01|          |A2    |A1    |       |     |
C1 |Jane  |female|1998.12.04|          |      |B1    |       |C2,C3|
C2 |John  |male  |1998.12.04|          |      |B1    |       |C1,C3|
C3 |George|male  |1998.12.04|          |      |B1    |  yes  |C1,C2|
C4 |Ann   |female|2003.02.04|          |      |B1    |       |     |
```

Listing 8: Example of Data File with Twins

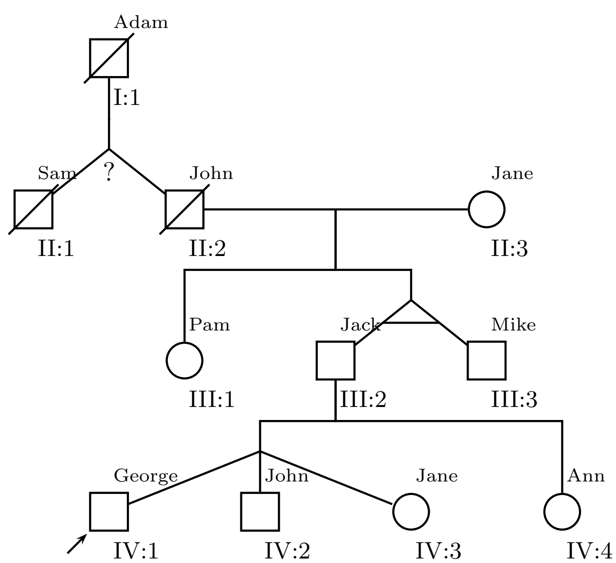
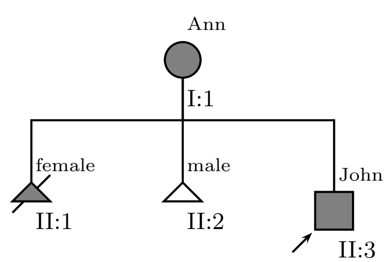


Figure 3: Example of a Pedigree with Twins (Data File from Listing 8)

```
Id |Name     |Sex   |DoB       |DoD       |Mother|Proband|Condition|Type
A0 |Ann      |female|1970.06.15|          |      |       |affected |
B1 |#abortion|female|1990.03.01|          |A0    |       |affected |
B2 |#abortion|male  |2000.10.10|          |A0    |       |         |sab
B3 |John     |male  |2002.12.01|          |A0    |yes    |affected |
```

Listing 9: Example of Data File with Abortions



I:1 Ann; born: 1970.06.15.

II:1 abortion; born: 1990.03.01.

II:2 abortion; born: 2000.10.10.

II:3 John; born: 2002.12.01.

Figure 4: Example of a Pedigree with Abortions (Data File from Listing 9)

```
Id |Name       |Sex   |Mother|Father|Proband|Type       |Comment
A0 |John       |male  |      |      |       |           |
B1 |James      |male  |      |A0    |       |           |
B1c|#childless |male  |      |B1    |       |infertile  |anospermia
B2 |Ann        |female|      |A0    |yes    |           |
B2c|#childless |      |B2    |      |       |           |
```

Listing 10: Example of Data File with Childlessness

Father: The `Id` of the childless male. If both `Mother` and `Father` columns are not empty, the entry describes the union between the `Father` and `Mother`. Of only `Mother` or `Father` is not empty, the entry describes the state of the corresponding person.

Type: This column might be either empty or have a keyword `infertile`. In the latter case the childlessness of the person or union is caused by a proven infertility.

Comment: The vaule of this column is shown under the childlessness symbol on the chart. Put there a short description of the cause of childlessness, like `anospermia` or `vasectomy`.

An example of a pedigree with childlessness is shown on Listing 10 and Figure 5.

4.6 Ordering Siblings and Marriage Partners

The generations in pedigrees are ordered in vertical direction, from up do down. How should we order the people on the same generation, i.e. siblings and marriage partners?

Usually two rules are used:

1. The siblings are ordered from the oldest on the left to the youngest to the right.

2. In marriage or other union the male is to the left, and the female is to the right.

However, the combination of these rules might lead to the situation when marriage lines intersect the parental lines. Therefore the rule 1 is usually implicitly modified:

1a. The are ordered from the oldest on the left to the youngest to the right. However, if a sibling's marriage is shown on a pedigree, this sibling is always the rightmost (male) or the leftmost (female).

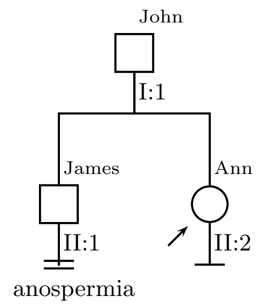


Figure 5: Example of a Pedigree with Childlessness (Data File from Listing 10)

The program follows these rules. It is enough to draw pedigrees in most cases. In particular, they always produce correct pedigrees if there is only one marriage shown. However, in complex cases these rules fail, as shown on Listing 11 and Figure 6. It is possible to extend the rules above to account for these cases, however we chose another solution: to provide a facility for the manual intervention in the sorting and ordering algorithm. For this purpose a special column `SortOrder` is used. It can have positive numbers greater than 1 or negative numbers smaller than -1. If the value of this column is positive, the corresponding person is moved to the left when sorting siblings and to the right when sorting marriage partners. If it is negative, the opposite sorting rule is applied (see Section 7 for more detailed discussion). Note that sibling sorting and marriage partners sorting must work in opposite directions, otherwise marriage lines intersect paternal lines.

Let us return to the pedigree on Listing 11. To improve Figure 6 we can either move Peter to the right or Lucy to the left. The first solution is shown on Listing 12 and Figure 7. The second is shown on Listing 13 and Figure 8.

Of course sometimes a pedigree cannot be drawn without self-intersections with any sorting of siblings. An example of such pedigree is shown on Listing 14 and Figure 9. Obviously no amount of shuffling the siblngs can help in his case.

If the program cannot avoid self-intersection of marriage lines and parental lines despite automatics sorting and manual intervention, as the last resort it creates a multi-segment marriage line, as shown on Figures 6 and 9.

```
Id |Name     |Sex   |DoB       |Father|Mother|Proband
A0 |John     |male  |1915.06.15|      |      |
B1 |Joan     |female|1940.03.02|A0    |      |
B2 |Jane     |female|1942.07.07|A0    |      |
B3 |Bill     |male  |1944.12.01|A0    |      |
B4 |Peter    |male  |1941.05.01|      |      |
C1 |Jack     |male  |1963.12.01|B4    |B2    |
C2 |Sam      |male  |1961.08.26|      |B1    |
C3 |Ann      |female|1965.11.12|      |B3    |
C4 |Lucy     |female|1965.12.11|      |      |
D1 |Mark     |male  |1989.06.21|C1    |C4    |yes
D2 |Dina     |female|1991.12.02|C1    |C4    |
```

Listing 11: A Data File with a Sorting Problem

```
Id |Name     |Sex   |DoB       |Father|Mother|Proband|SortOrder
A0 |John     |male  |1915.06.15|      |      |       |
B1 |Joan     |female|1940.03.02|A0    |      |       |
B2 |Jane     |female|1942.07.07|A0    |      |       |
B3 |Bill     |male  |1944.12.01|A0    |      |       |
B4 |Peter    |male  |1941.05.01|      |      |       |  3
C1 |Jack     |male  |1963.12.01|B4    |B2    |       |
C2 |Sam      |male  |1961.08.26|      |B1    |       |
C3 |Ann      |female|1965.11.12|      |B3    |       |
C4 |Lucy     |female|1965.12.11|      |      |       |
D1 |Mark     |male  |1989.06.21|C1    |C4    |yes    |
D2 |Dina     |female|1991.12.02|C1    |C4    |       |
```

Listing 12: First Solution to the Problem in Listing 11

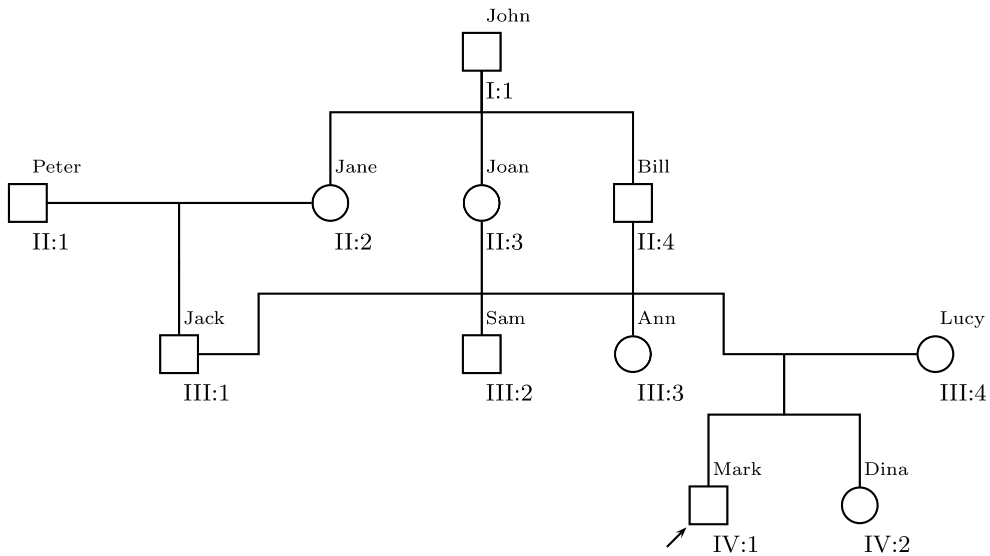


Figure 6: Pedigree from Listing 12

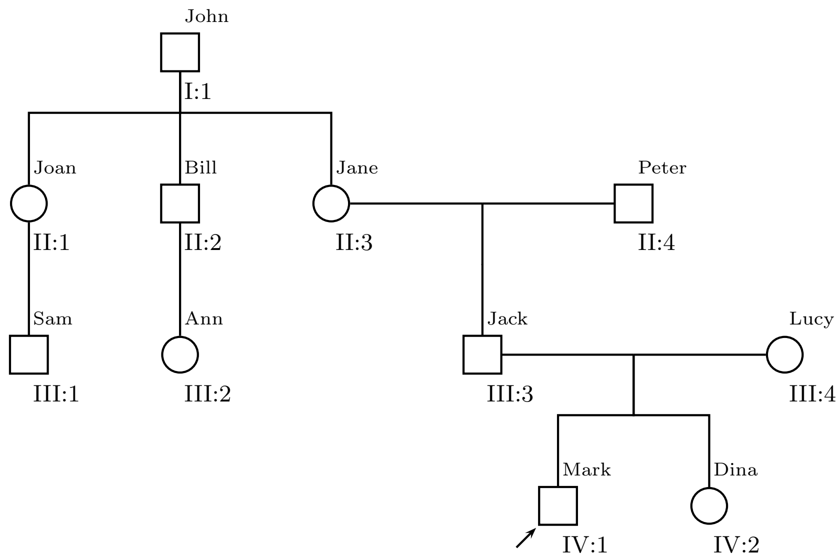


Figure 7: Pedigree from Listing 12

```
Id |Name     |Sex   |DoB       |Father|Mother|Proband|SortOrder
A0 |John     |male  |1915.06.15|      |      |       |
B1 |Joan     |female|1940.03.02|A0    |      |       |
B2 |Jane     |female|1942.07.07|A0    |      |       |
B3 |Bill     |male  |1944.12.01|A0    |      |       |
B4 |Peter    |male  |1941.05.01|      |      |       |
C1 |Jack     |male  |1963.12.01|B4    |B2    |       |
C2 |Sam      |male  |1961.08.26|      |B1    |       |
C3 |Ann      |female|1965.11.12|      |B3    |       |
C4 |Lucy     |female|1965.12.11|      |      |       | -3
D1 |Mark     |male  |1989.06.21|C1    |C4    |yes    |
D2 |Dina     |female|1991.12.02|C1    |C4    |       |
```

Listing 13: Second Solution to the Problem in Listing 11

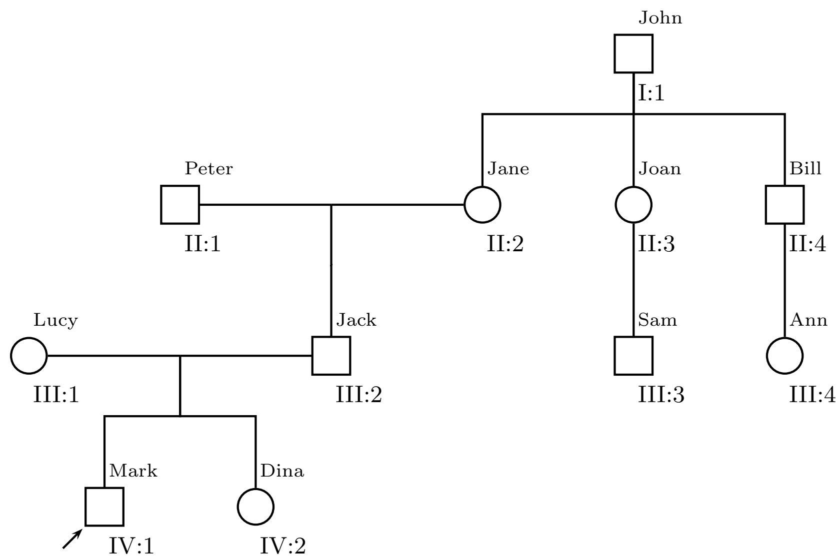


Figure 8: Pedigree from Listing 13

```
Id |Name     |Sex   |DoB       |Father|Mother|Proband
A0 |John     |male  |1915.06.15|      |      |
B1 |Sam      |male  |1935.12.04|A0    |      |
B2 |Ann      |female|1937.03.02|A0    |      |
C1 |Paul     |male  |1952.10.03|B1    |      |
F1 |Scott    |male  |1912.02.01|      |      |
G1 |Simon    |male  |1934.09.17|F1    |      |
G2 |Sarah    |female|1936.12.19|F1    |      |
H1 |Lola     |female|1960.04.13|G2    |      |
K1 |Jim      |male  |1962.11.05|G1    |B2    |
M1 |Jane     |female|1917.02.13|      |      |
P1 |Simon    |male  |1935.10.04|      | M1   |
R1 |Pam      |female|1964.02.05|P1    |      |
X1 |James    |male  |1988.07.12|K1    |R1    |yes
```

Listing 14: A Pedigree with Unavoidable Self-Intersections

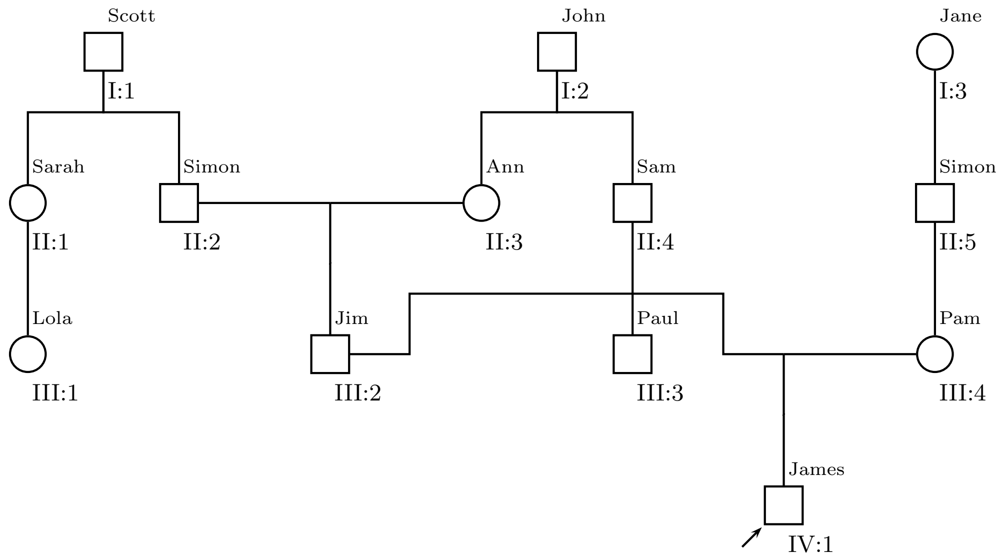


Figure 9: Pedigree from Listing 14

```
Id |Name     |Sex   |Father|Mother|Proband|DoB
A0 |Jane     |female|      |      |       |1908.12.12
B1 |John     |male  |      |A0    |       |1936.12.15
B2 |Ann      |female|      |A0    |       |1934.04.17
B3 |Samantha |female|      |A0    |       |1932.12.03
B4 |Nancy    |female|      |A0    |       |1928.01.05
C1 |Mary     |female|      |B2    | yes   |1955.08.26
C2 |Paul     |male  |      |B3    |       |1964.05.07
C3 |Jane     |female|      |B4    |       |1950.11.03
D1 |Jack     |male  |B1    |C1    |       |1975.07.01
D2 |Laura    |female|C2    |C3    |       |1974.09.05
```

Listing 15: A Pedigree with Consanguinic Unions

4.7 Consanguinic Unions

Consanguinic unions present a technical problem for the program (see the discussion in Section 8). Therefore the support of consanguinicity is experimental for this release.

There is a number of limitations for consanguinic unions in the data file at present. First, the consanguinic unions should not in the direct lineage of the proband or the person from which the pedigree starts. In many cases this limitation can eliminated by using `-s` option (see Section 4.1) to choose a different starting point for the pedigree. Second, the children of consanguinic unions might appear not centerd on the charts. An example of a pedigree with consanguinic marriages is shown on Listing 15, and the corresponding chart is shown on Figure 10. The drawbacks of the program are evident from the positions of Laura nad Jack on these charts.

4.8 Language-Dependent Keywords

At present the program pedigree can work with English and Russian languages. As discussed in Section 3.5, the language options chooses *both* the languages of input and output files. It is easy to add new languages to the scheme by expanding the library `Pedigree::Language.pm` in the distribution.

The English language is the default. Moreover, if the Russian option is chosen, English keywords are still recognized in the input file.

The English and Russian keywords are listed in Table 1. Note that some keywords have variants; they are listed in the table as well.

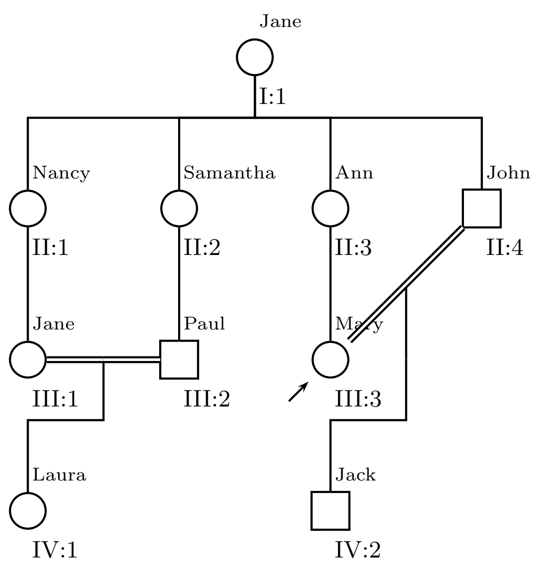


Figure 10: Pedigree from Listing 15

English keyword	English variants	Russian keywords
Field Names		
Id		Идент
Name		ФИО
Sex		Пол
DoB		Рожд
DoD		Умер
Mother		Мать
Father		Отец
Proband		Пробанд
Condition		Состояние
Comment		Комментарий
Type		Тип
Twins		Близнецы
SortOrder	Sort	ПорядокСортировки, Сорт
Field Values		
male		муж, м
female		жен, ж
unknown		неизв, неизвестно
yes		да
no		нет
normal		норм, здоров
obligatory	obligat	облигат
asymptomatic	asymp	асимп
affected	affect	больн, болен
infertile		бесплодн
sab		выкидыш
monozygotic	monzygot	монозиготн, монозиг, однояйцев
qzygotic	qzygot, ?	?
Special Names		
#abortion		#аборт
#childless		#бездетн

Table 1: Keywords in Different Languages

Part II
Algorithm Description

5 Introduction

This part is intended for advanced users and is not neccessary for runnuing the program.

The problem of nicely typesetting graphs is one of the classical problems in the Computer Science [4]. One of the earliest algorithms here is the classical algorithm for layered rooted trees by Reingold and Tilford [4, § 3.1]. This algorithm was implemented by `PSTricks` [3]. However, many pedigrees are not trees [2]. If we consider a subset of pedigrees where inbreeding is absent, the pedigrees become trees. However, even in this case the the tree is not necessary layered, as can be seen from Figure 1. Therefore a new approach generalizing Reingold-Tilford algorithm is necessary. This approach is based on the analysis of the structure of pedigrees and is sketched in the remainder of this manual.

6 Main Algorithm

A pedigree consists of nodes (vertices), connected by lines (edges). If there is no inbreeding, the graph is acyclic. There are two kinds of nodes in the graph: person nodes (squares and circles on Figures 1 and 2) and *marriage nodes*, which are nameless on the figures. We will use the notation "male spouse-female spouse" for such nodes, so the marriage nodes on Figure 1 are I:1-I:2, I:3-I:4 and II:2-II:3. A node has a *precedessor* and *children.* A marriage node does not have a precedessor, but has *male spouse* and *female spouse* (it is customary to put male spouses to the left and female spouses to the right on pedigrees). Any node has a *downward tree* of its children, grandchildren etc. The downward tree may be empty.

Any node in an acyclic graph can be a root. However, in layered trees there is a special root: the one that has no precedessor. Similarly we will call a *local root* a node that has no predecessor. All marriage nodes are local roots. Some person nodes can be local roots as well.

Let us first discuss the case where cobnsanguinic marriages are absent. In this case a pedigree is a tree.

The proposed algorithm is recursive and starts from a local root. Strictly speaking, it can start from any local root, but medical pedigrees have a special person: *proband,* the person who was the first to be examined by genetic specialists (the proband is shown by an arrow drawn near the node on Figures 1 and 2). Therefore it makes sense to start from the local root which has proband in its downward tree.

If this local root is a person node, the pedigree is the layered tree, and Reingold-Tilford algorithm is sufficient. Therefore we should consider only the

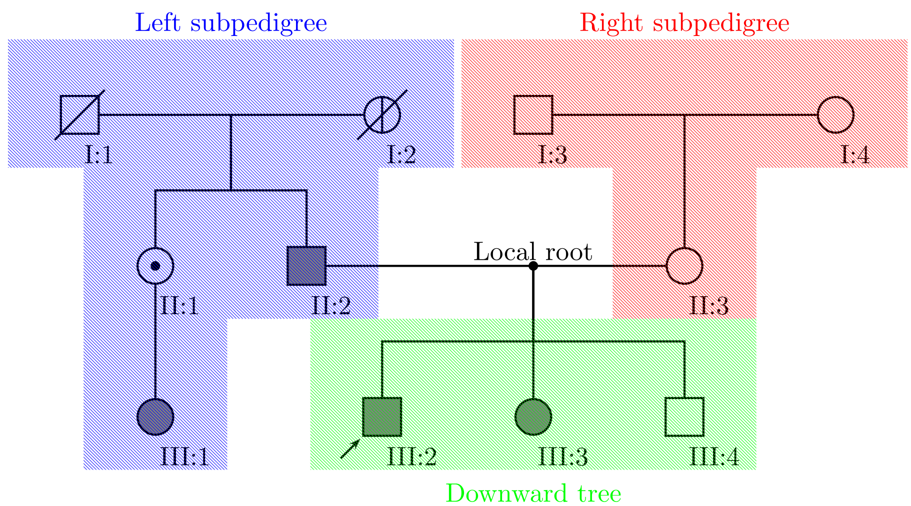


Figure 11: Subpedigrees and Downward Tree

case when the local root is a marriage node. In this case we can typeset the downward tree using Reingold-Tilford algorithm. The spouses do not belong to this tree. However, each of them belongs to each own subpedigree. We will call them *left subpedigree* and *right subpedigree*. We recursively apply our algorithm to typeset left and right subpedigrees. Then we move the left subpedigree to the right and right subpedigree to the left as far as we can without intersection between them and the downward tree.

This process is shown on Figure 11. Obviously this algorithm converges and leads to typesetting the pedigree without intersections between the subtrees and subpedigrees.

7 Algorithm for Sorting Siblings and Marriage Partners

When we create a marriage node, we want to put the male to the left and the female to the right. When we then sort siblings, we want this male to be the rightmost, and the female to be the leftmost. To do so, we assign to each node the special quantity `SortOrder`. Initially all nodes have `SortOrder` equal to zero, unless specifically set by the user in the input file (see Section 4.6). Then we use the following rules:

1. When creating the the marriage node:

(a) If both spouses have equal `SortOrder` field, the male goes to the left, the female goes to the right.

(b) Otherwise, the spouse with greater `SortOrder` goes to the left.

(c) If `SortOrder` of a spouse is 0, we set it to 1 (the spouse on the left) or -1 (the spouse on the right).

2. When sorting siblings:

   (a) The sibling with smaller `SortOrder` goes to the left.

   (b) If both siblings have the same `SortOrder`, the oldest one goes to the left.

8 Modifications for Consangunic Unions

Consanguinic unions present a problem for the described algorithm, because pedigrees with them are no longer trees (see Figure 10).

In this release of the program we use the following hack. The direct lineage of the proband (or, more generally, the starting node) may have both mothers and fathers in the pedigree because they share genes from the starting node. If any other person has both mother and father in the chart, his or her parents both shared their genes with the starting node. Therefore they formed a consanguinic union. In this case the children of this node appear in two subtrees: their mother's and their father's.

We delete them from one of the subtrees (the one with lower generation number), connect their parents with a double line (consanguinic union) and put the descent line from the middle of the union to them.

There are two problems with this hack (see Section 4.7): the children of consanguinic unions are not centered on the diagaram, and the hack fails if the starting node itself is a descendant of a consanguinic union.

Probably the next releases will employ better algorithms for consanguinic unions.

9 Conclusion

The algorithm seems to be efficient and producing nicely typeset pedigrees. Since the input file format is simple, it may be used by the people without special skills in LaTeX. On the other hand, the TeX files produces are easy to understand and edit manually if the need arises.

10 Acknowledgements

The authors are grateful to Herbert Voß for help with `PSTricks` code. The support of TeX User Group is gratefully acknowledged. One of the authors (LA) was supported by Russian Foundation for Fundamental Research (travel grant 06-04-58811), Russian Federation President Council for Grants Supporting Young Scientists and Flagship Science Schools (grant MD-4245.2006.7)

References

[1] Robin L. Bennett, Kathryn A. Steinhaus, Stefanie B. Uhrich, Corrine K. O'Sullivan, Robert G. Resta, Debra Lochner-Doyle, Dorene S. Markei, Victoria Vincent, and Jan Hamanishi. Recommendations for standardized human pedigree nomenclature. *Am. J. Hum. Genet.*, 56(3):745–752, 1995.

[2] Boris Veytsman and Leila Akhmadeeva. *Creating Medical Pedigrees with PSTricks and LaTeX*, July 2007. `http://ctan.tug.org/tex-archive/graphics/pstricks/contrib/pedigree/pst-pdgr`.

[3] Timothy Van Zandt. *PSTricks: PostScript Macros for Generic TeX*, July 2007. `http://ctan.tug.org/tex-archive/graphics/pstricks/base/doc`.

[4] Giuseppe Di Battista, Peter Eades, Roberto Tamassia, and Ioannis G. Tollis. *Graph Drawing: Algortihms for the Visualization of Graphs.* An Alan R. Apt Book. Prentice Hall, New Jersey, 1999.